CLONING, EXPRESSION AND CHARACTERIZATION OF THE SPG4 GENE RESPONSIBLE FOR THE MOST COMMON FORM OF AUTOSOMAL DOMINANT SPASTIC PARAPLEGIA

ABSTRACT OF THE DISCLOSURE

The invention concerns the identification and characterization of the SPG4 gene encoding spastin, and some mutations thereof responsible for the most frequent form of autosomal dominant familial spastic paraplegia, to the cloning and characterization of its cDNA and the corresponding polypeptides. The invention also concerns vectors, transformed cells and transgenic animals as well as diagnostic methods and kits, and methods for selecting a chemical or biological compound capable of directly or indirectly interacting with said polypeptide.